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|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |  |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | (43) International Publication Date: 21 September 2000 (21.09.00) |
| (21) International Application Number: PCT/AU00/00193<br>(22) International Filing Date: 17 March 2000 (17.03.00)<br>(30) Priority Data:<br>60/124,864                      17 March 1999 (17.03.99)                      US<br>9908190.3                      9 April 1999 (09.04.99)                      GB<br>(71) Applicant (for all designated States except US): WOMEN'S AND CHILDREN'S HOSPITAL [AU/AU]; 72 King William Road, North Adelaide, S.A. 5006 (AU).<br>(72) Inventors; and<br>(75) Inventors/Applicants (for US only): MEIKLE, Peter, John [AU/AU]; 31 Minnamurra Drive, Redwood Park, S.A. 5097 (AU). HOPWOOD, John, Joseph [AU/AU]; 2 Monarto Court, Stonyfell, S.A. 5066 (AU). WINCHESTER, Bryan, Gordon [GB/GB]; 9 Lake Gardens, Ham, Richmond, Surrey YW10 7SJ (GB).<br>(74) Agent: SPRUSON & FERGUSON; G.P.O. Box 3898, Sydney, NSW 2001 (AU). |  | (81) Designated States: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CR, CU, CZ, DE, DK, DM, DZ, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, TZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG).<br><br><b>Published</b><br><i>With international search report.</i> |                                                                   |
| (54) Title: DIAGNOSIS OF LYSOSOMAL STORAGE DISORDERS USING SAPOSINS AND OTHER MARKERS                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |  |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |                                                                   |
| (57) Abstract                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |  |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |                                                                   |
| <p>The invention provides methods of diagnosing or monitoring lysosomal storage disorders based on detecting levels of saposins, LAMPs and/or <math>\alpha</math>-glucosidase in patient sample. Elevated levels of saposins and/or LAMPs are indicative of a disorder. Elevated levels of <math>\alpha</math> glucosidase are indicative of some types of lysosomal storage disorders and decreased levels of <math>\alpha</math> glucosidase are indicative of other types of lysosomal storage disorder. In some methods, the profile of elevation of different saposins, LAMPs and <math>\alpha</math> glucosidase allows distinction between different types of lysosomal storage disorder.</p>                                                                                                                                                                    |  |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |                                                                   |